SYMPOSIUM ON USHER'S SYNDROME

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SYMPOSIUM ON USHER'S SYNDROME

Washington, D. C.
April 19, 1973

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FOREWORD

Gallaudet College is glad to have had a part in bringing together individuals with relevant information on the needs of people with Usher's syndrome. The increasing incidence of children in schools for the deaf with hereditary deafness indicates a corresponding proportionate increase in the number of deaf children with retinitis pigmentosa.

Only a few students in any single school for the deaf will have this additional disability. Moreover, the fact that severe visual impairment for academic purposes usually does not manifest itself until the middle or late teens deceptively masks the needs of affected individuals during their school years. The result is a growing number of bewildered young adults and families with Usher's syndrome victims who have no understanding of the problems and little recourse to agencies able to provide guidance or appropriate services.

Fortunately, this situation can be rectified and the needs of these individuals can be met by schools for the deaf during regular school years. Reliable and inexpensive screening for retinitis pigmentosa can be accomplished by schools for the deaf with existing personnel. Program adjustments for these students can be instituted without great

disruption to existing programs.

As a result of this conference Gallaudet College will proceed with a number of activities recommended in this report. These recommendations include actions which schools for the deaf and other agencies should pursue in order to begin to meet the needs of Usher's syndrome victims.

Gallaudet College itself is prepared to examine its own programs to determine how it can be of further assistance in areas such as continued acceptance of college-level students with Usher's syndrome and possibly in the need for special courses for professional personnel interested in working with deaf-blind youths and adults.

We wish to express our appreciation to the participants of this long needed symposium. Especially we wish to acknowledge the valuable efforts of Mr. Arthur Roehrig, a Gallaudet College graduate from Wisconsin with Usher's syndrome, who continues to identify and interest new professional resources in the problems of deaf people with visual problems.

Albert T. Pimentel, Director
Public Service Programs

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OVERVIEW OF USHER'S SYNDROME: CONGENITAL DEAFNESS AND PROGRESSIVE LOSS OF VISION

McCay Vernon, Ph. D.

Usher's syndrome, a genetic condition causing congenital deafness and progressive loss of vision, was discovered in 1858. Unfortunately it took over a century, one hundred and fifteen years to be exact, to progress from the discovery of the disease to the first coordinated multidisciplinary effort to do anything constructive about its prevention and the rehabilitation of those who have it. It is almost unbelievable that a condition which causes three to six percent of deaf children to become blind is still ignored by professionals in deafness.

To illustrate the situation it is helpful to provide some documentation. First, no school or college for the deaf in the United States has an adequate screening program for the identification of those who have Usher's syndrome. The schools that have routine visual acuity screening do not include the necessary additional ophthalmological procedures to identify the condition nor are the screening examinations conducted often enough to assure early diagnosis. Furthermore, few, if any, schools have staff members including physicians, psychologists,

audiologists, teachers, or administrators familiar with the disease. For example, at Gallaudet College, where students with the disease have it in a sufficiently advanced stage to permit identification, who has identified the most cases? It is not the medical, audiological or other professional staff but a student, Art Roehrig. Mr. Roehrig has no ophthalmoscope, no visual field testing equipment, no Snellan Chart, and no dark adaptation room, yet it is he who has identified most of the Gallaudet cases who have been diagnosed. Mr. Roehrig has done this because, as a person with the disease, he cares. In addition, Mr. Roehrig knows some of the symptoms of the condition such as "tunnel vision," night blindness, and vertigo. This is not altogether a criticism of Gallaudet because if secondary schools had appropriate visual screening, all those with the disease would be identified prior to enrollment at Gallaudet.

Another illustration of the way the problem is ignored is the policy of the Department of Health, Education, and Welfare which financed the research that led to the development of a prevention program for the disease (Grinker, 1969; Vernon, 1969). Instead of implementing the prevention plan or adequately disseminating information about the disease in order that it might be prevented, the Department has spent millions of dollars to establish regional centers for the care and diagnosis of deaf-blind persons. Half of deaf-blindness is due to Usher's

syndrome (Vernon, 1969). The point of all this is to dramatize the responsibility to alter the inertia of the one hundred and fifteen years from the time Usher's syndrome was identified until now.

What is Usher's Syndrome?

Usher's syndrome is a recessive genetic condition the major symptoms of which are congenital deafness and a progressive loss of vision leading to eventual blindness (Kloepfer, Laguaite, and McLaurin, 1966). Usually, the patient first notices difficulty seeing at night during his early teens or twenties. An observable narrowing of the visual field then occurs. At about this time the diagnosis of retinitis pigmentosa is generally made.

Two major professional problems come to light at the time of diagnosis. First, few of those who diagnose the retinitis pigmentosa connect it with the deafness. Consequently, it is not identified as Usher's syndrome. The second difficulty is that in addition to not diagnosing the presence of Usher's syndrome the person "making the diagnosis" is often unaware of the genetics of the disease or even the fact that it is a recessive inherited condition. Thus, the genetic counseling, screening, and follow-up which should occur does not and the disease is spread instead of being prevented (Nance, 1973; Vernon, 1969).

Several other aspects of Usher's syndrome need description. Along with the deafness and loss of vision there may be, but are not necessarily, some additional disorders (McLeod, McConnell, Sweeney, Cooper, and Nance, 1971). Among these are mental retardation, vertigo, psychosis, loss of sense of smell, abnormal EEG's, and epilepsy (Vernon, 1969).

In discussing the nature of Usher's syndrome one anecdotal finding is of interest. Following the publication of a paper on Usher's syndrome in 1969 I received letters and referrals from people who read the article and realized that they had the disease. Many of these persons were bright successful individuals who in some cases were only hard-of-hearing and partially sighted and in other cases had retained much more vision until an older age than previous research had reported (Vernon, 1969). The reason for this may be that previous studies included samples which had been institutionalized, i.e., they were in centers for the deaf-blind, schools for the deaf, mental hospitals, etc. Thus, these studies yielded patients more severely involved than would have been the case had a randomly selected sample of cases been surveyed. This suggests that there may be a significant number of persons with Usher's syndrome in whom residual vision and sight are retained longer than previous findings had indicated. Perhaps there are also more cases than had been thought in which only vision and sight

are affected and the other involvements such as vertigo are absent.

Some of the problem grows out of the confusion between Usher's syndrome (retinitis pigmentosa, deafness, and cerebellar ataxia), Lawrence Moon Biedl syndrome (retinitis pigmentosa, deafness, retardation, obesity, polydactyly, and hypogonadism), Alstrom's syndrome (retinitis pigmentosa, deafness, obesity, and diabetes) and Refsum's disease (retinitis pigmentosa, deafness, obesity, ichthyosis, and polyneuritis). A reason for this confusion is that all of these diseases include the two basic components of deafness and retinitis pigmentosa which are part of Usher's syndrome.

A hopeful note relative to treatment and Usher's syndrome is that Refsum's disease has been found to be due to an identified enzymatic defect. This has led to the development of a dietary supplement which may prove effective in halting the progression of Refsum's disease and a reversing of some of the neurologic abnormalities (Nance, 1973). Usher's syndrome is closely enough related that similar treatment breakthroughs may occur with it. Dr. Nance (1973) also stated that the enzymatic defect in Refsum's disease could be detected in the fluid surrounding the unborn fetus. If Usher's syndrome could be similarly detected, it would be a major step forward in prevention. One question which arises is, has the data on Refsum's disease been applied to research on the treat-

ment and prevention of Usher's syndrome?

Diagnosis

At present Usher's syndrome is generally identified by ophthalmoscopic examination, i.e., the doctor looks into the eye with a light focused through a magnifying lens. The trouble with this method of examination is that it does not identify the disease until it is well advanced and the patient is in his teens or twenties. This is much too late for the most efficient genetic counseling. For example, these patients have often married and started a family without knowing they have the condition. It is also past the time that would be ideal for designing appropriate educational and rehabilitative approaches.

Effective early diagnosis requires the techniques listed below. The use of the last three would be ideal for screening a high risk population in programs for deaf children (Bergsma, 1973).

1. Electroretinography
2. Electroculography
3. Dark adaptation recording
4. Visual field testing
5. Ophthalmoscopy

The point of early diagnosis is that it makes possible prevention and rehabilitation through genetic counseling, screening of relatives, and provision of appropriate rehabilitative approaches.

Prevention

Prevention of a significant amount of Usher's syndrome is possible for a very unique reason. Normally one would think that such a disease which only occurs in three per one hundred thousand people would not be feasible to screen for due to the high costs involved. However, the unique reason screening for Usher's syndrome can be effective is the centralized availability and identifiable nature of the high risk population, who are the congenitally deaf. Their centralization makes it relatively easy to provide the ophthalmologic and perhaps auditory screening required for early detection ... detection which leads to the identification of carriers and to genetic counseling which can prevent the spread of the disease. The logistics of this require some exploration.

The first condition leading to the centralization of deaf children is that before they go to school most of them have audiological tests in one of the three hundred and fifty or so speech and hearing clinics in the United States and Canada (Craig, 1973). Approximately two hundred and seventy-seven of these are in medical schools or colleges (Craig, 1973). The remaining sixty plus are in schools for the deaf or private facilities (Craig, 1973). Thus, the high risk population is identified, centralized and available in these clinics at pre-school age.

Once deaf children become school age over two-thirds of them are in one of the one hundred and sixty-six residential or large day schools (Craig, 1973). Many of the remaining one-third are in day classes in large metropolitan areas (Craig, 1973).

The high risk population can be even further narrowed down because among deaf children those with known exogenous causes of deafness (up to fifty percent) can be eliminated, often by a simple check of existing school records and family pedigrees.

In brief, this means that through a relatively inexpensive screening procedure a yield rate of six to thirty percent will result for a disease which has a prevalence of three per one-hundred thousand in the general population (Kloepfer et al., 1966; Vernon, 1969). This is a remarkable and fortuitous circumstance we are totally failing to capitalize upon.

The second phase of the prevention program would involve screening of relatives of the deaf children found to have the disease or to be carriers of it. A third dimension would be the screening of relatives of identified cases of retinitis pigmentosa without deafness. Usher's syndrome is far more common among them than in the general population (Vernon, 1969).

Based on all these data the following five point prevention program is submitted.

I. Educate professional persons (medical and allied specialists) who serve deaf or blind children about Usher's syndrome. This includes at a very minimum, otologists, ophthalmologists, pediatricians, audiologists, psychologists, teachers, and administrators serving programs for the deaf and hard-of-hearing.

II. The second step in prevention is the screening of all congenitally deaf children with an opthalmologic battery including ophthalmoscopic examination, visual field test, and dark adaptation measures.

III. The third step in prevention is the administration of an audiologic testing battery consisting of Bekesy (sweep and fixed tracings), SISI, tests of vestibular functioning, loudness and variable speed discrimination testing. The theory behind this is that audiologic patterns will be found in cases of Usher's syndrome that may also be present in carriers of the disease (Kloepfer, et al., 1966).

IV. Relatives of identified cases of Usher's syndrome should be given full opthalmological, audiological, and perhaps biochemical screening.

V. Genetic counseling should be provided all identified cases and carriers as an immediate follow-up.

Conclusions

At present most deaf people with Usher's syndrome get no real rehabilitative service. When young they generally function as deaf people. As blindness gradually overtakes them they are left isolated and unprepared to cope.

The field of deafness faces the responsibility of undoing the inertia of the last one hundred and fifteen years and providing something constructive relative to Usher's syndrome. This responsibility includes implementing a prevention program, a rehabilitation program, and a research effort geared to curing or arresting the visual loss associated with the disease. Furthermore, there is a need to provide some reasonable estimate of the costs and an identification of the appropriate sources of funding for such efforts.

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GENETIC ASPECTS OF USHER'S SYNDROME

Walter E. Nance, M. D., Ph. D.

Progress in medicine during the past few decades has led to the elimination or alleviation of so many of the important environmental causes of disability and disease that conditions in which an abnormality in host is responsible for or contributes to the disability will assume increasing importance in the future. This trend is particularly striking in relation to the causes of deafness. Within the past few years there have been dramatic advances in the management of premature infants, the treatment of ear infections and the prevention of rubella and Rh incompatibility which promise to reduce if not eliminate some of these important "environmental" causes of deafness. Geneticists estimate that at the present time about fifty percent of profound deafness is genetically determined. In view of the factors we have just discussed, this proportion will undoubtedly rise in the future.

The most important insight that has emerged from research on hereditary deafness during the past decade is the recognition that genetic deafness is not a single disease entity but a collection of many different disorders which have one symptom in common, namely, hearing loss.

Considering the extraordinary complexity of the human organ of hearing, this insight should perhaps have come as no surprise. The precise interaction of literally hundreds of genes must be required to provide the genetic instructions that are necessary for the formation of a normal ear and a defect in any one of many different genes can therefore cause deafness.

Usher's syndrome is only one of several recognized forms of retinitis pigmentosa. In contrast to the dominant and X-linked forms of retinitis pigmentosa, Usher's syndrome is transmitted as an autosomal recessive trait. This means that the disease is caused by the possession of a pair of abnormal genes, one inherited from each parent. The parents each carry a single abnormal gene but are clinically normal. At present, there is no reliable method of detecting the carrier state (i.e., individuals who possess a single dose of the abnormal gene) for Usher's syndrome. Even in families where both parents are carriers of the gene for Usher's syndrome, the chances that each child will be affected are only one in four. This explains why physicians often fail to recognize the hereditary nature of the deafness in families where Usher's syndrome has occurred. Unless the physician is trained in genetics or particularly alert to the possibility that he may be dealing with a hereditary form of deafness he may not even consider this possibility unless there are two or more deaf children in the family. However,

the average size of human families has become so small that in about eighty to eighty-five percent of Usher's syndrome families there is only one affected subject. The condition is no less inherited or genetically determined in these families but by chance none of the patient's brothers and sisters are affected. It is unfortunate that the possibility of a genetic form of deafness is so often not considered in these isolated cases because, at least in the case of Usher's syndrome, the special diagnostic tests reviewed by Dr. Bergsma can often permit an accurate diagnosis to be made long before the onset of the progressive loss of vision that typically begins during the second decade of life. How the early knowledge of impending loss of vision would influence the educational plans that should be made for an affected individual is a major challenge for those involved in special education for the multiply handicapped. However, I can think of no better example of the potential importance of making a specific diagnosis of the particular type of genetic deafness that an affected individual has.

Usher's syndrome can vary greatly in age of onset, severity and speed of progression. One explanation for this variation is that mutations of several different genes may mimic one another in that they all may cause retinitis pigmentosa and hearing loss -- the two cardinal features of Usher's syndrome. For example, Refsum's disease is a condition in which retinitis pigmentosa and hearing loss are associated

with ataxia, heart disease and cutaneous manifestations in some patients. The specific enzymatic defect in Refsum's disease has been characterized and an effective form of therapy has been developed. If we only had a comparable understanding of the biochemical abnormality in Usher's syndrome, it might be possible to devise a specific treatment for that disorder as well.

Another possible cause for clinical variation in Usher's syndrome is the presence in the population of many different mutations involving the same gene. In the case of the two gene pairs which determine the human hemoglobin molecule, for example, several dozen different mutations of the same gene have been identified which may have widely different clinical effects. As we learn more about Usher's syndrome we should expect a comparable degree of genetic heterogeneity to emerge. One practical consequence of these facts is that the progress and severity of the disease in other affected members of the same family is perhaps the best guide in predicting the future course that a patient will follow.

Even in the absence of a complete understanding of the biochemical defect in Usher's syndrome or the availability of a specific treatment, a great deal can be accomplished through genetic counseling. Counseling should be provided for affected individuals as well as their normal parents.

In general, genetic counselors have often failed to provide counseling for affected individuals at a time before marriage when counseling would be most relevant to them. An important point to emphasize both for patients and their carrier parents is the ubiquity of abnormal genes in the general population. It is estimated that everyone carries about two to seven abnormal recessive genes, but it is only if we carry a double dose of the same abnormal gene that a recessive genetic disease results. Consequently, the load of abnormal recessive genes is a burden that all society bears, and the social stigma that was formerly attached to genetic diseases of all types can be seen to be a reflection of ignorance. These facts also emphasize the futility of attempting to eliminate abnormal genes from the general population by eugenic means. If we passed a law prohibiting anyone who carried an abnormal gene from reproducing, no one could have children, since we all carry abnormal genes whether we know it or not. The strategy we should adopt in genetic counseling is to try to identify high risk couples, that is to say parents who both carry the same recessive gene, so that we can provide them with accurate data concerning their risk of bearing affected offspring upon which they can then make an informed decision concerning their family plans.

What effect, we might ask could the widespread application of genetic counseling have in reducing the frequency of Usher's syndrome? Suppose every case of Usher's syndrome that occurred in the population

were correctly identified by a wide-scale screening program and that genetic counseling were provided for the parents of every affected child that was discovered. Suppose further that the counseling was "one hundred percent effective" in the sense that every couple who was counseled decided to have no more children because of their twenty-five percent recurrence risk. What effect would this have on the incidence of the disease in future generations? It may come as a surprise to many to learn that a "one hundred percent effective" system of this type would reduce the incidence of the disease by only about fifteen to twenty percent and the program would have to be continued for many future generations thereafter for any substantial further decrease in the disease incidence to occur. The reason why the effects of counseling would not be more dramatic is that for every affected individual in the population there are literally hundreds of carriers that we have no way of detecting at present. If we wait until an affected child is born before we provide counseling (and there is no other choice if we cannot detect the carrier state), we can only prevent recurrences of the disease in a family. As we saw earlier, in only about fifteen to twenty percent of families with Usher's syndrome is there more than one affected child, and this provides a rough measure of the proportion of cases that could be prevented by counseling. In order to achieve a greater reduction in the disease incidence through counseling it would be necessary to develop a reliable

method of carrier detection so that couples who were at risk of having affected offspring could be identified before they actually have an affected child.

Counseling for individuals who are affected with Usher's syndrome is generally more optimistic than they expect. Unless the marriage partner is also affected with Usher's syndrome or is a carrier of the gene for Usher's syndrome there is virtually no risk that their children will have Usher's syndrome. Since the incidence of carriers in the general population is only about one in two to three hundred, the risk that an individual with Usher's syndrome will have an affected child is very small if he marries an unrelated person with normal hearing. Even if the marriage partner is also deaf, the chance of a child with Usher's syndrome will be very low unless the spouse also has Usher's syndrome in which case all their children will be affected.

In my opinion, it is not necessary to justify genetic counseling in terms of its effect on the overall incidence of a genetic disorder. I feel that the real value of counseling lies in the psychological benefits that the family can derive from a clear understanding of the cause and prognosis of the disease and the medical and educational benefits the patient may obtain because of early diagnosis. The opportunity that genetic counseling provides to prevent the recurrence of the disease

in families who know better than anyone else what the total impact of Usher's syndrome can be is a final benefit that cannot be overemphasized.

Several existing resources might be utilized to implement a program aimed at the early detection of Usher's syndrome. The Office of Demographic Studies at Gallaudet College presently collects data on approximately forty-five thousand deaf children each year. Information is already available from the survey which suggests that at least four percent of deaf children have an associated severe visual handicap. It seems quite likely that many of these children may have undiagnosed Usher's syndrome, a possibility that could be explored in future surveys. A second approach to the diagnosis of hereditary deafness that I have been developing deals with an attempt to link the pedigrees of living children with deafness to a large sample of pedigrees of deaf couples that were obtained by E. A. Fay before the turn of the century. In this way it might be possible to identify some cases of deafness as being hereditary and conceivably to suspect a particular type of deafness, such as Usher's syndrome, because of the presence of affected relatives.

A final recent development which has revolutionized genetic counseling should be mentioned. For an increasing number of specific diseases, prenatal detection is now possible. This means that high risk couples (if they wish) can monitor their future pregnancies and complete

their families with normal children by electing to have a selective abortion performed if an abnormal fetus is detected. Unfortunately, no technique is currently available for diagnosing Usher's syndrome prenatally, but it is possible that techniques of this type may be developed in the near future.

Question:

Wiedenmayer: Is it true that only women transmit the autosomal recessive gene that causes Usher's syndrome? What are the chances of my grandchildren having it?

Nance: There are several genetically distinct forms of retinitis pigmentosa that either show different patterns of inheritance or different associated findings. One common form of retinitis pigmentosa is caused by an abnormal gene on the X chromosome, and shows what we refer to as a "sex-linked pattern of inheritance." Normal females have two X chromosomes while males have only a single X and a very much smaller Y chromosome. Because males have only a single X, any abnormal gene carried on that chromosome will be fully expressed. Females, on the other hand, ordinarily would require a double dose of an abnormal recessive X-linked gene to show full expression of the trait. This means that half of the sons of a carrier female would be expected to be affected, and half of her daughters would be carriers. In contrast, none of the sons of an affected male would be affected but all of his daughters would

be carriers and would have a risk of bearing affected sons.

Usher's syndrome is another form of retinitis pigmentosa which is distinguished by the associated hearing loss and has an entirely different inheritance pattern. In this case the disease is caused by possession of two copies of an abnormal recessive gene that is carried on one of the twenty-two pairs (we don't know which pair) of non-sex chromosomes or autosomes. Consequently, we would expect all of the unaffected children of an affected parent, regardless of their sex, to be carriers of a single abnormal gene. Their chance of having an affected child, however, would be very low because for this to happen their spouse would also have to be a carrier of the same abnormal gene. Unless the spouse is a close relative, the chance that he or she would be a carrier of the same recessive gene would be very low, something in the range of one chance in two hundred or less, in the case of Usher's syndrome.

OPHTHALMOLOGIC ASPECTS OF USHER'S SYNDROME

Donald R. Bergsma, M.D.

Usher's syndrome is the genetic disease resulting in deafness and retinitis pigmentosa, which produces a slowly progressive loss of night and side vision. The condition is inherited in autosomal recessive fashion as described by Dr. Nance.[1,2] Although hearing difficulty frequently occurs during adulthood in patients who have retinitis pigmentosa, Usher's syndrome generally refers to the combination of profound, congenital deafness and retinitis pigmentosa. This paper will focus on the clinical course of the eye disease, methods of detection and prevention, and factors to be considered in rehabilitation of patients with Usher's syndrome.

At present, Usher's syndrome is distinguishable from the more common forms of retinitis pigmentosa only by the presence of profound, congenital deafness. Therefore, as an ophthalmologist, I shall describe the clinical course of retinitis pigmentosa without continuously specifying that we are referring to Usher's syndrome.

Retinitis pigmentosa is a slowly progressive degeneration of the retina and pigment epithelium (the layer next to the neural retina) which is characterized by:

(1) loss of night vision

(2) loss of side vision

(3) pigmentary clumping

(4) loss of the electroretinogram (ERG) which is an electrical potential normally produced by the retina and pigment epithelium in response to light

Estimates of the frequency of Usher's syndrome vary widely. As Dr. Vernon[3,4] and Dr. Nance[1,2] have indicated, the frequency is very low in the general population but probably occurs among approximately four per cent of the profound congenitally deaf population. In fact, the incidence may be higher because the signs and symptoms of retinitis pigmentosa may not be present or detected until after school age.

The onset of visual difficulty may occur from childhood to middle age. Most commonly the night blindness begins many years before the loss of side (peripheral) vision is disabling. The central (reading) vision is generally maintained for many years after extensive loss of peripheral vision has occurred. Note that no exact time frame is given because age of onset varies widely.

The symptoms of retinitis pigmentosa are (1) night blindness (poor dark adaptation) which is usually present to some extent from childhood but usually does not bring the patient to the doctor, (2) poor side vision (visual field loss) which is usually detectable by the teens but frequently the patient is unaware of the problem until it is fairly advanced, (3) poor central vision

which is usually not a problem until late in life, although exceptional cases occur earlier. Moreover, loss of side vision makes central vision less useful than normal even when the central vision is normal. Note that these symptoms often will be manifest in the deaf individual indirectly, for example, missing hand or gesture communication in groups or difficulty with school work due to decreased reading speed.

There are several tests for retinitis pigmentosa. The diagnosis is usually detected by observation of pigment clumping and narrowing of blood vessels inside the eye. Such detection depends heavily on dilation of the pupil with medication, because not all areas of the retina are equally involved and the signs may be missed if dilation is not used.

A second test, dark adaptation, measures the dimmest light that can be seen after entering a dark room. Normally there is a rapid increase in ability to see the dim light (increasing sensitivity, decreasing threshold) during the first twenty minutes in the dark. Retinitis pigmentosa produces slow and incomplete dark adaptation. Since not all areas of the retina are affected, mild cases occasionally could go undetected unless the test is administered so as to check specified areas of the retina. Therefore, screening for mild cases would be cumbersome with this test, but as will be noted later, it may be of some advantage not to detect mild cases at too early an age.

Visual field testing detects a loss of peripheral vision which typically begins in the mid-periphery producing a donut shaped area of poor vision

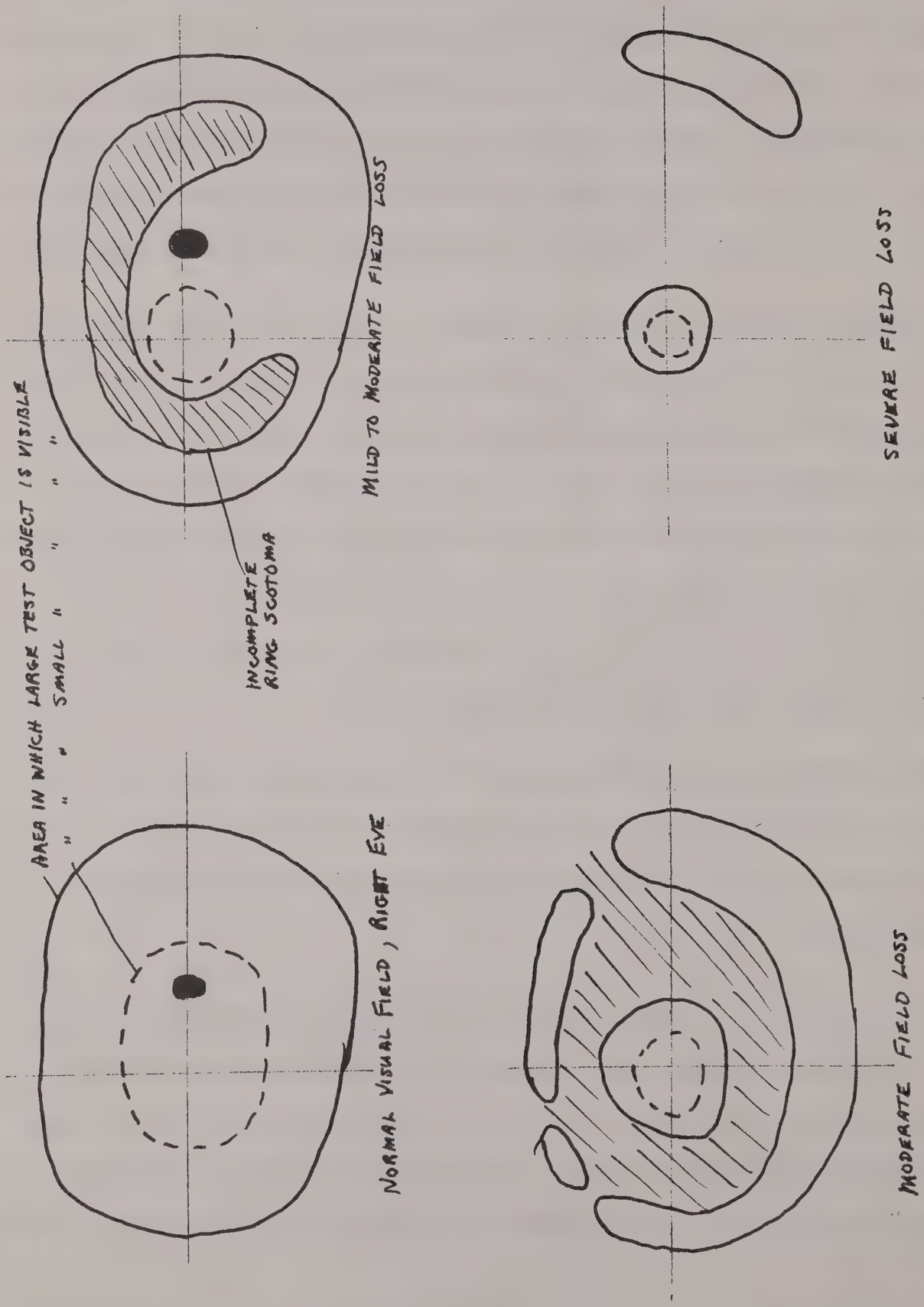
("ring scotoma"). Later in severe cases only the very central vision and frequently a peripheral island of vision remain, see Figure 1. The ability to detect mild or moderate loss of peripheral vision depends heavily on the testing conditions. Severe loss of peripheral vision is detectable by many methods. Therefore, as with other tests, detection of retinitis pigmentosa in deaf children requires careful definition of the methods of detection.

A fourth test for retinitis pigmentosa is the electroretinogram (ERG). The ERG measures the electrical potential produced by the retina and the pigment epithelium in response to light. It is diminished or absent in retinitis pigmentosa. This test is extremely helpful in detecting early cases when such information is desirable for definitive diagnosis of unusual cases or for genetic counseling. At present the ERG, available only at major eye care centers, should not be performed unless there are specific indications and is not a good mass screening tool.

Fluorescein angiography is a special technique using fluorescein dye to visualize defects in the pigment epithelium in retinitis pigmentosa. It is necessary only in unusual circumstances and is not appropriate as a screening test.

The prognosis of a patient must be individualized depending on the specific findings in each case. Although the gene for Usher's syndrome apparently differs from that for retinitis pigmentosa, the clinical experience in both conditions indicates extremely wide variation of age of onset, severity and rate of progression. If an older family member has been affected,

FIGURE 1


AREA IN WHICH LARGE TEST OBJECT IS VISIBLE
" " " SMALL " " " "
INCOMPLETE RING SCOTOMA
MILD TO MODERATE FIELD LOSS
SEVERE FIELD LOSS
NORMAL VISUAL FIELD, RIGHT EYE
MODERATE FIELD LOSS

this may serve as a general but not precise guide as to the expected course of the younger patient. The general prognosis is one of slow progression over many years. Typical stages are (1) poor dark adaptation: childhood, disabling only at night, (2) poor peripheral vision: onset during teens, not disabling until advanced, a process which takes many years but usually is disabling to some degree by middle age, and (3) poor central vision: usually a problem in old age but can occur early. Dr. Vernon has mentioned that some patients with Usher's syndrome sometimes develop neurological difficulty or endocrine abnormalities.[4] While this possibility should be considered by the physician, it seems unwise to discuss this with the patient unless neurologic or endocrine abnormalities are already present because this usually does not occur and may in fact represent confusion with another genetic disease, such as the Hallgren and Alström syndromes.[5]

Rehabilitation of visually handicapped individuals often begins when they reach the level of legal blindness, which is defined as visual acuity of 20/200 or worse in the better eye or less than 20 degrees of horizontal field. Many agency criteria differ, however, and it is extremely important to remember that the double handicap of profound deafness and poor vision is much greater than the sum of the single handicaps. Much compensation for deafness is through sight and vice versa. The patient with Usher's syndrome will, therefore, require rehabilitation at a somewhat earlier stage of visual loss than will the hearing individual with isolated retinitis pigmentosa. Moreover, it probably is advisable , despite the psychological burden it places on the patient, to have deaf-blind rehabilitation training before the

patient is totally handicapped visually to avoid the huge communication difficulties which arise if one waits until after blindness occurs in Usher's syndrome. (Generally patients facing progressive loss of vision are unwilling to undergo rehabilitation until after the handicap has incapacitated them. I am not, however, advocating extremely early detection nor rehabilitation before the patient with Usher's syndrome has some handicaps visually.)

At present, rehabilitation for loss of night and peripheral vision is best accomplished with mobility training (long white cane technique, etc.). Rehabilitation for patients with loss of field to the extent that reading and visual hand signal communication are impaired requires tactile communication training. Furthermore, there are many techniques which make daily activity easier for the trained deaf-blind or deaf-partially sighted individual.

As must be apparent by now, there is no known effective treatment for preventing visual loss in retinitis pigmentosa nor for restoring retinal function once it is lost. However, some patients are aided by visual aids depending on the status of their central and peripheral vision and on their specific visual needs. In addition, a large percentage of patients with retinitis pigmentosa eventually develop cataracts and, if the cataract is advanced, its removal may result in improved vision for the patient despite partial loss of retinal function from retinitis pigmentosa. However, the timing of a cataract extraction is a matter of judgment which must be

decided between the patient and his ophthalmologist with many factors in mind. I would caution against removing the cataract at too early a stage.

Prevention of the disease can be accomplished to a small degree by genetic counseling. However, as Dr. Nance has pointed out, because we are unable to detect carriers by clinical testing (except, of course, affected patients have certain relatives who are either obligate carriers or have a probability of being carriers *a priori*), most new cases will arise as the first affected member of a family.[1] On the other hand, it is important that patients with Usher's syndrome be identified and given genetic counseling before the age of marriage, particularly since they run a high risk of meeting friends carrying the same genetic defect at schools for the deaf.

This leads finally to a discussion of plans for detection of patients with Usher's syndrome. At first glance it might appear that the techniques which I have described above should be adapted to screening patients with non exogenous causes of deafness at very early ages. However, there are several factors which make extremely early detection undesirable. First, some of the techniques are less accurate or more difficult to apply at an early age. Second, some mild cases would be missed producing false reassurance. Third, other cases would be detected long before rehabilitation was necessary or desirable and before any sort of meaningful prediction could be made as to when significant handicaps could be anticipated or what the ultimate visual fate of the patients would be. This situation would produce extreme anxieties,

and unnecessarily so. Finally children with Usher's syndrome should learn as much visually as possible as long as their vision is adequate. Therefore, it is my considered judgment that screening programs for Usher's syndrome would best be directed at the appropriate deaf populations at the high school level. This would allow time for proper occupational decisions to be made. Of course individuals who develop signs or symptoms of the disease at earlier or later ages can be referred for ophthalmologic examination without screening programs.

The logistics and specific techniques of effective screening programs are complicated. Care must be taken to utilize techniques which do not produce excessive numbers of false positive or false negative results. Back up personnel, facilities and resources must be available to properly work up and counsel those who are identified as possibly being abnormal on the basis of the screening examinations. Therefore, I would strongly advise that experience be gained via small, well planned pilot screening programs before large ones are undertaken. Simpler screening programs to check visual acuity and perhaps fundus examinations and intraocular pressure measurements are, of course, highly desirable for all deaf children at intervals starting from pre-school age in order to detect correctable visual problems.

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COPING WITH THE DISCOVERY OF USHER'S SYNDROME

Arthur A. Roehrig

To be told that you have Usher's syndrome is really depressing and miserable, but not as traumatic as some people might think. It is because there are many things that might help you to cope with Usher's syndrome before you discover it. For instance, the loss of night vision is the first thing that makes you realize that your vision is deteriorating. The loss of side vision is next.

As you may already know, Usher's syndrome is the inherited combination of congenital deafness and progressive blindness, which is caused by retinitis pigmentosa. Congenital deafness is accepted easily because the sense of hearing is never experienced. The progressive blindness is usually accepted, although more slowly. It could be traumatic, however, if a person is directly told that he has retinitis pigmentosa which is a major cause of blindness.

There are three levels of awareness of deteriorating vision in the victim of Usher's syndrome:

1. unconscious awareness - when a person realizes his inability

to see at night

2. semi-conscious awareness - when he reaches the point where he notices his eyes are continually deteriorating

3. conscious awareness - when he is directly told that he will be blind

Now, let me tell you about how I became unconsciously aware of my visual limitations and how I felt about it. When I was ten years old and a pupil at St. John's School for the Deaf in Milwaukee, Wisconsin, some friends and I were playing a game of hide and seek. It was the first time I played the game at night, and it was the first event that made me realize I had difficulty seeing at night. When we started playing the game, I saw all of the boys running to many places where they thought they could not be seen. I thought I could do this and started running. All of a sudden, I ran into trees and tripped over things, such as rocks and bushes. However, what I did that night did not bother me because I thought all the boys were running into things also. But when the game was over, we went into a well-lighted building. My supervisor, a nun, and all the boys suddenly stared at me and the supervisor said to me, "What happened to your face, arms and clothes?" I became puzzled and did not know what she meant. So when I went to a bathroom, I looked in a mirror and then at the other boys' faces, arms, and clothes. Although I began realizing my inability to see at night, I

thought all the boys also couldn't see well but had avoided the obstacles because they had memorized all they saw during the day.

Two months later, a night in winter, all the boys and I went out and sledded in the snow. We were on the top of a hill and waited for turns to sled down. There were a lot of trees and a barn down beyond the foot of the hill. At that time I could hardly see what the boys were doing; however, I went ahead and sledded. I sledded several times and enjoyed it without difficulty. Then I had an accident in an area which should have been clear. I ran into something, and it fell on top of me. I felt the object and suddenly realized it was my teacher, a nun.

At that moment I was embarrassed and upset, and knew that my teacher would blow her top. But surprisingly, she did not. She was very kind and understanding. When she took me to a place where I could see her, she asked in a nice manner why I hit her with my sled, and I nervously said that I had not meant to do it and wanted to apologize. She, however, said it was unnecessary for me to make an apology. I asked why. She finally told me that the accident happened because I could not see at night as normally as the boys at the school. At first I did not believe that, but a few moments later, I remembered the night when I played hide and seek and had bruises and scars all over my face and arms while the other boys did not. I became unconsciously aware of

my visual impairment and accepted it easily. The reason I was able to accept it easily was because I had been allowed to continue to do many things that normally sighted people do.

I remained unconsciously aware of my visual impairment for a long time, even after I found out that I have tunnel vision, until one afternoon in a classroom at the Maryland School for the Deaf in Frederick, Maryland. At that time, I was employed as a second-year teacher in the academic year 1970-71. I had been reading a newspaper and when I rubbed my left eye while continuing to read, I suddenly noticed that I could not see with my right eye as before. My vision was wavy. It made me realize that my eyes would continue deteriorating. Because of this new realization and because I had been unconsciously aware of the problem for a long time, I, at this point, became semi-consciously aware of my visual impairment.

When I began my third and final year of teaching in the fall of 1971, my vision was still becoming worse. When I entered my classroom, I knew that it was more and more difficult to read on a blackboard and to understand what my students said. I started thinking what would happen to my eyes in the next few years. Of course, I became depressed and worried about it, so I decided to stop teaching and to spend as much time as possible seeing several eye doctors. The more

I knew about what would happen to my eyes, the better I could prepare myself adequately for my future.

About three and one-half months later, I finally entered as an in-patient at the National Institutes of Health in Bethesda, Maryland. I stayed there for about a month for a complete head-to-toe examination. During my stay at the institute, my social worker, Mrs. Schumann, and my ophthalmologist, Dr. Donald Bergsma, worked very closely with me. One day, Mrs. Schumann and I started talking about my future. At that time, she, in a friendly and nice way, tried to talk me into thinking a little more about the things that blind people are encouraged to learn, such as braille and cane-mobility, and I then became consciously and hopelessly aware of what would happen to my eyes in the next few years -- I mean blindness. The night following my talk with Mrs. Schumann, I was naturally depressed and cried all night because I knew I would be blind some day even before Dr. Bergsma explained to me about my progressive blindness.

The fact that I became aware of my deteriorating vision through a gradual process enabled me to accept this handicap with less trauma than I may have experienced otherwise. Psychologically, several other things helped me accept the reality of my progressive blindness.

An awareness that there are individual differences among all

people enabled me to accept my own differences. I was born with Usher's syndrome. It was neither my fault nor my parents'. It is important for me to know this. It is impossible to know why some of us are born deaf or blind just as it is impossible to know why some of us are born black and others,white. Every person is unique. This makes him different from every other individual. Yet every individual is beautiful and is equally as important as anyone else. This means that I am as important as any person of yesterday, today, or tomorrow, including President Nixon. This is exactly what all my family members, the staff members and students at my alma maters, and all the people I know have taught me.

As long as a person is alive he has a right to be known, understood, and loved. He must be given every opportunity to experience and understand everything he is able to see, hear, touch, smell, and taste. These senses are the best gifts a person receives in his lifetime.

Understanding that there are differences among all people was helpful to me in learning to understand and accept myself. Being honest with myself and totally aware of who I am and what I have has helped me develop my self concept. I have learned my strengths and weaknesses, my abilities and limitations. In addition to being honest with myself, I feel it is important to be honest with other people by letting them also

know my strong and weak points so that they are able to be comfortable when they are with me. My self concept includes understanding the differences between things I am responsible for and things which I am not responsible for. For instance, I am not responsible for becoming congenitally deaf and progressively blind, but I am responsible for knowing what to do with my double handicap.

In closing, I really feel that every victim of Usher's syndrome is an important person, and special plans must be made for him. I admit that I owe my life to Jesus, God, my family members, the staff members and students and many other people I know for teaching me to be just plain Arthur Roehrig and nothing else.

TUNNEL VISION AMONG THE HEARING IMPAIRED: SOME PERSONAL EXPERIENCES

Joseph Wiedenmayer

Bing, bang, clump! I had driven across a concrete safety isle in Milan, Italy. Endeavoring to make a left turn, I had not seen the raised concrete platform along side of my car. Fortunately, I stopped in time to miss an oncoming bus. My passengers were scared, so I said, "Gosh, these Italians don't care where they pour their concrete." That wisecrack relieved the tension.

That shakeup occurred in 1950 and I soon forgot about it until some weeks later when I had an eye infection. I went to a famous Italian ophthalmologist and surgeon for examination and treatment. He cauterized my right eye and then he said to me, "You need eyeglasses but you also have RP." He explained that RP is a progressive eye disease which constricts the visual fields; that is, limits or eliminates peripheral (side) vision. "It is not operable, nor can it be arrested or cured by medicinal means, and, being progressive, it can result in total blindness in some cases," he added.

That was the first time I knew I had RP, characterized by "tunnel

vision." In previous years, as far back as 1943, ophthalmologists had mentioned that there was some constriction of my visual fields but I was never told specifically what the problem was. Although I had been cautioned about driving, I gave little thought to the matter. Certainly, it did not occur to me that the accidents I had had prior to the one in Milan were probably caused by tunnel vision. I assumed it was just carelessness on my part. Certainly they were not the result of my deafness which was congenital and progressive.

Since then, I learned that some congenitally deaf or hard-of-hearing people with visual problems have RP but don't know it. This combination is sometimes referred to as "Usher's syndrome." Normally hearing people are also afflicted by this progressive eye disease.

As I look back over the years, I recall specific incidents and accidents and I realize that nearly everything I hit or got hit by was from the side, not head on. During World War II, at my first post, the United States Embassy in Montevideo, Uruguay, I was walking across a street when all of a sudden I was knocked down by a running horse and wagon. I was shocked, bruised, and dirtied, but not seriously hurt. The horse didn't step on me. I was only late for work.

Another time while I was on leave in the United States, I drove smack into the side of a truck, badly damaging both vehicles. But

aside from bumping into things and knocking over drinks, one of my most embarrassing experiences was when I was at a formal reception at the U. S. Embassy in Rome in 1949. As I entered the room, I spotted the Ambassador and walked briskly up to him and extended my hand to shake his. I missed and bumped into a waiter carrying a large platter of juicy canapes which he immediately dumped all over the Ambassador's blue suit. Some guests laughed but it wasn't funny. One of the Embassy officers accused me of being intoxicated. I wasn't, of course. I told him only that I was clumsy sometimes.

Over the years, even after I knew about my eye disease, I never told anyone in the Foreign Service about it nor the extent of my deafness -- not even the State Department Medical Examiners. Reading an eye chart or printed words does not reveal tunnel vision. My visual acuity or forward vision was also deteriorating. Finally, towards the end of my career, I became legally blind. Nevertheless, I tried to be a good "cover-upper" of my problems to protect my career. When I walked I took a friend's arm -- not for help, of course, but rather as a friendly gesture. As for driving, I always had a chauffeur because it was more elegant -- certainly not because I couldn't drive.

The extent and nature of my blindness and deafness was not discovered by the U. S. State Department Medical Branch until several

months before I retired "for age" in 1965, and that resulted only because I had asked for an eye and ear examination at the Bethesda Naval Hospital in the hope that surgery, at government expense, might be helpful.

Once again I was told that corrective eye surgery was not possible. In fact, the doctor predicted that I would become completely blind in four to six years due to advanced tunnel vision combined with posterior subcapsular cataracts. He was wrong. At the Naval Hospital I did have ear surgery (stapedectomy) but there was no improvement at all in my hearing so I have remained unable to hear any sounds without a powerful hearing aid in one ear though not in the other.

Finally in 1971 I found an ophthalmologist at Johns Hopkins Hospital willing to operate on my worst eye. He removed the cataract. Normally a common operation, it was complicated in my case due to RP. It is not known which condition was the major cause of my blindness. Now I can see better with long vision lenses but I am still legally blind. No doctor in the United States or abroad ever offered a glimmer of hope for even arresting my tunnel vision. There is one hope today for the future -- RESEARCH.

Everyone with impaired vision, regardless of the extent and nature of it, should have his peripheral vision examined occasionally. It should also be incumbent upon every ophthalmologist to tell the patient specifi-

cally about eye diseases he may have.

Every school for the deaf should have all students examined for tunnel vision because loss of peripheral vision restricts one's ability to read manual language or lipread effectively. A student with this double handicap needs special recognition by the teachers.

A FAMILY MEMBER'S REACTION TO USHER'S SYNDROME

Lil E. Browning

The first time I can remember hearing the words retinitis pigmentosa was when I was around fifteen years old.

My mother had been seeing an eye-ear-nose-and-throat specialist for many years for prescription lens needs. Around 1953, she was advised to consult an eye specialist. In Waco, Texas, at that time population of around fifty thousand, there was only one ophthalmologist. Mom was seen at six-month intervals for a couple of years. All the while, she became more and more agitated with the physician's services because her vision continued to blur increasingly and because the doctor would not respond to her requests for more powerful lens.

In retrospect, I feel sure the ophthalmologist became frustrated in his attempts to communicate with her by pencil and pad due to my mother's poor comprehension of medical terms and the fact that she had a great deal of difficulty reading. (No Flair felt-tip pens then!) He was leery of giving her the full impact of his diagnosis, perhaps because of his limited ability to provide supportive counseling.

And, in all probability, he wished she were not his patient.

I say this because one day after school, Mom, very angry and hurt, met me at the door telling me I must go with her to see the eye doctor. She had been to see him that day and he had brushed her questions and requests off with his note to her that she was not to return to his office again unless she were accompanied by a hearing person.

Mom's pride was deeply scarred. Until that time, she thought, she had operated pretty self-sufficiently and independently. (I said "she thought" because in subsequent years I learned from my paternal grandmother and my father's sister -- both of whom lived in Waco -- that most of my parents' dealings with businesses and physicians were monitored via phone calls by my father's relatives.)

Mom and I marched to the bus stop and she directed me to the ophthalmologist's office. The receptionist said my mother had no appointment but left to consult with the doctor. Mom and I were ushered into his office where he was to see us on many other occasions.

The ophthalmologist told me to relay to Mother that she had retinitis pigmentosa. He briefly discussed the condition with me and advised me not to explain the prognosis to my mother. He did encourage me to fully relate his explanation to my father. It seemed to me that

my mother had me ask the doctor a hundred times that visit whether or not she would become blind. His standard reply was, "Tell her that I do not know." Aside, the doctor told me blindness was the usual result of retinitis pigmentosa, but that since he had no way of predicting how long my mother would live, he refused to commit himself with a direct answer to Mother's question.

Mother's frustrations, unhappiness, and periods of depression are marked quite deeply in the memories of my mid-teen years. How my father took all her outbursts and somehow managed to stabilize his functioning is still a mystery to me.

In 1958, I was to move away from home for good -- three years, summers included, attending a university in another city, followed by marriage and residence in Fort Worth.

While I was in college Mom wrote that the eye specialist had at last admitted she would soon be blind; she had bilateral cataracts. Mom was aware that cataracts could be removed surgically and was very angry when the ophthalmologist refused to operate on her eyes. He would not tell her that the retinitis pigmentosa could have progressed to the stage that, even if the cataracts were removed, she would still be blind.

Everyone around my mother was miserable! Mother wanted to go to a physician who would operate and remove the cataracts; she and Dad would pay monthly on the surgeon's fee and the hospital bill. The rest of the family sympathized and bemoaned that a person would be weighted with a double sensory handicap. Which one of us would dare to dismiss the physician's advice and try to explain what retinitis pigmentosa -- my mother called it "my eye disease" -- was and its documented effects?

Since I was not available, other relatives began to serve as intermediary resources between the ophthalmologist and my mother. The Commission for the Blind accepted my mother as a client and agreed to underwrite the cost of the cataract removal. Those involved wondered what might happen to Mother if indeed the retinitis pigmentosa had done its damage and Mother would have to face blindness following surgery. The Commission furnished a hearing, blind woman to teach Mother braille.

Mother insisted that others could see again after cataract surgery and that she would, too. She startled the braille teacher one day when I was home when she threw the braille reader across the room and yelled at me to tell the teacher and her dog to leave and never come back!

My aunt had bitter words with the doctor; finally he relented and

agreed to operate on the eye that had been the lesser of the two ravaged by retinitis pigmentosa prior to the development of the cataracts.

The doctor's instructions that Mother should not move her head at all during the surgery and the first few days of recuperation were interpreted by Mom to mean for six weeks. She wanted so desperately to see again! No amount of cajoling and stating that she need not hold her head and neck so rigidly for so long had any effect. She would do nothing to jeopardize the possibility of regained vision. Consequently, for many months following surgery my mother suffered from strained muscles in the upper back and neck and frequent, severe headaches. But, she could see! Within six months of the first cataract surgery, Mom convinced the doctor to remove the cataract from the other eye.

For the following ten years, Mom enjoyed fairly good vision. She could never again be independent in moving about outside the lot around our house -- sunlight blinded her and she could not see in the total darkness of night -- but she memorized the terrain of the land and was able to go to the wash house and hang clothes on the line by herself.

In the mid-sixties, a hearing-impaired friend of my mother's suffered severe visual problems and mastered braille. He was able to convince Mom that she, too, should learn braille. Her vision had limited her to headlines of newspaper articles and titles of magazine

articles. Every Sunday afternoon for about two years, my parents welcomed the Draughons; Mom and Mr. Draughon rehearsing the meaning of the raised configurations of dots and Dad and Mrs. Draughon catching up on the latest gossip.

Braille has furnished Mom with countless hours of information and entertainment. Her vision continues to diminish; she complains of sunbursts in her eyes, of moving and flashing bits of light when she moves quickly or is startled. Her ability to see seems related to her emotions, state of mind, and curiosity.

She is still able to prepare all the meals, but Dad now fingerspells the recipe instructions in Mom's hands, does all the measuring, sets the oven and burner controls, and tells Mom when the recommended time has elapsed. Often, however, by her refined sense of smell, Mom is able to detect the stage of doneness of the food and reminds my father to check the food.

Everything in the apartment is in a certain place. Quickly Mother memorizes locations and items. Her most common complaint is of people who forget to completely close cabinet or closet doors.

She has been hypertensive for as long as I can remember and a constant worrier. Few things are completely satisfactory to her and she often complains, but then she follows with a smile.

AMERICAN COUNCIL OF THE BLIND

Joseph Wiedenmayer

At the time of the Symposium efforts were being made to establish a committee within the American Council of the Blind which would be a national, non-profit organization for deaf-blind adults. This has now been accomplished.

The Committee for the Deaf-Blind held its first meeting in July, 1973. There are six members on the committee -- half of whom are deaf-blind themselves. Two Symposium participants are committee members -- Mr. Robert Smithdas and Mr. Joseph Wiedenmayer who acts as Chairman. Deaf-blind people with problems should appeal to this committee for assistance. Improving the image of deaf-blind people and providing a forum through which deaf-blind people, who have completed their training, can exchange ideas are functions of this committee within the American Council of the Blind.

BUREAU OF EDUCATION FOR THE HANDICAPPED

Robert Dantona

The Bureau of Education for the Handicapped is the only federal program concerned with the education of handicapped children. The rubella epidemic of 1964-65 stimulated Congress to pass legislation which provides services to deaf-blind people. Under provisions of P. L. 91-230, the Education of the Handicapped Act, ten Regional Centers for Deaf-Blind Children were established. Each is headed by a regional coordinator and provides services for five to seven states. Their services are offered "to children who have both auditory and visual impairments, the combination of which causes such severe communication and other developmental and educational problems that they cannot properly be accommodated in special education programs either for the hearing-handicapped child or for the visually-handicapped child."

Through these regional centers approximately three thousand and six hundred deaf-blind children have been located. The centers provide diagnostic and educational assessment services to these deaf-blind children as well as individual training and counseling for the children and their families. Other functions of these centers are development of

training programs for personnel working with deaf-blind children and dissemination of information and materials pertinent to the area of services to the deaf-blind. Information should be made available to personnel at these centers about early identification of retinitis pigmentosa.

HELEN KELLER MEMORIAL FUND

Edward C. Carney

"Hearing Conservation and Work for the Deaf" has been designated by the Lions International Foundation as a major objective of the organization. In Alexandria, Virginia, a Lions Club of deaf men has established the Helen Keller Memorial Fund. The purpose of this Fund is to accumulate financial support for projects in the area of deafness. Presently there is about $12,000 in the Helen Keller Memorial Fund.

Traditionally supportive of the blind, the Lions International Foundation is now concerned with developing films and printed materials which would provide information about deafness. Possibly they could be encouraged to expand this area of interest to include the needs of deaf-blind people.

NATIONAL CENTER FOR DEAF-BLIND YOUTHS AND ADULTS

Louis Bettica

The National Center for Deaf-Blind Youths and Adults was formulated under President Johnson in 1967 and became a reality in 1969. Presently it is housed in a temporary facility which can handle only eighteen to twenty people at a time; however, a new building is planned which should be able to accommodate fifty persons.

Since the National Center can provide comprehensive services to a limited number of individuals at its temporary facility, the Center employs a strict definition of deaf-blindness for eligibility for admission to the Center. Applicants must be reasonably ambulatory and able to care for themselves. Good language ability is not necessary, for about one-third of the deaf-blind population has been bypassed by schools for the deaf and for the blind.

Counseling services for deaf-blind clients begin before they enter the National Center and continue even after they have left. Only forty-two to forty-four people have left the Center. Fifty-two percent of these are now remuneratively employed. Many of these people have

retinitis pigmentosa.

Three regional offices have been established to assist various communities in providing services for deaf-blind people. There is a great need for orientation of personnel working in this field around the country.

NATIONAL RETINITIS PIGMENTOSA FOUNDATION

Joseph Wiedenmayer

The National Retinitis Pigmentosa Foundation, located in the Rolling Park Building, 8331 Mindale Circle, Baltimore, Maryland 21207, was founded in 1971. The primary function of this organization is to raise funds for research on this hereditary, genetic eye disease. A secondary function is distribution of information on retinitis pigmentosa.

Through television advertising the National Retinitis Pigmentosa Foundation helped raise $975,000 to set up a laboratory at the Massachusetts Eye and Ear Infirmary for research on retinitis pigmentosa. Hopefully, in time, concentrated research will result in a cure for this disease.

The National Retinitis Pigmentosa Foundation is compiling a confidential national registry of youths and adults with retinitis pigmentosa. The purposes of the registry are to determine the incidence of this disease among hearing people and among deaf and hard-of-hearing people and to inform such people and their relatives of significant developments in research on this disease. Anyone who has retinitis pigmentosa is encouraged to write to Mr. Wiedenmayer in order to be included in the registry and in order to receive information about retinitis pigmentosa.

VOCATIONAL REHABILITATION ADMINISTRATION
WASHINGTON, D. C.

Constance Gant

Washington, D. C. at one time had the only state program for deaf-blind people. At the program's inception five years ago sixty-five blind people who could also be categorized as hard-of-hearing or deaf were located on the blind register that the Vocational Rehabilitation Administration had been keeping. Since that time referrals have been received from speech and hearing clinics, the Columbia Lighthouse for the Blind, schools, Public Assistance, ear and eye doctors, and hearing aid dealers. Through this program, evaluation, training, and job placement for deaf-blind people has resulted in employment as key punch operators, power sewing machine operators, packers, rehabilitation counselors, maids, homemakers, and wake-up service operators.

In addition to vocational placement, goals of the program for the deaf-blind in Washington, D. C. have included the following:

1. Establishing a recreational program for the deaf-blind so they will be able to have some opportunity for social life.

2. Finding resource people who are able to talk with

and assist clients with problems such as alcoholism or mental illness.

3. Locating community agencies and volunteers who would accompany deaf-blind people to medical appointments.

4. Teaching communication techniques to families of deaf-blind clients.

5. Identifying deaf people with visual problems.

In considering rehabilitation of the deaf-blind, there are two distinct groups -- those who become blind first and have verbal skills and those who are congenitally deaf and, for all practical purposes, are nonverbal. The cost for the rehabilitation of the congenitally deaf person who later becomes blind is almost twice that of a blind person who becomes deaf. This is because of the training involved in overcoming the language handicap in addition to braille and mobility training.

Many years ago, when Mary Switzer was Administrator of the Social and Rehabilitation Service, the Department of Health, Education, and Welfare issued a mandatory vocational rehabilitation regulation that required all legally blind persons be given hearing examinations. There is no mandatory regulation saying that all persons with congenital hearing problems must have eye examinations. This should be rectified. Another area of limited service to deaf and deaf-blind people is provision of interpreter services. Many times vocational rehabilitation agencies

do not have sufficient funds to pay for interpreters for medical and other consultations hearing-impaired people may need.

RECOMMENDATIONS

Identification and Screening Needs

1. The diagnosis of Usher's syndrome can be medically accomplished with confidence by the time a child is eight years old. Early identification programs should be initiated in schools for the deaf and for the blind. Superintendent Joe Shinpaugh of the Virginia School for the Deaf and the Blind should present this recommendation to the Conference of Executives of the American Schools for the Deaf and to the appropriate organization representing schools for the blind.

2. The Office of Demographic Studies at Gallaudet College should solicit in their annual survey of schools for the deaf as much information as possible on the incidence of deaf children with severe visual impairments; particularly valuable would be information on screening activities used in schools for visual impairments.

3. Competent medical authorities in the field should develop guides for screening procedures, such as dark room testing techniques, for use by schools for the deaf. The National Eye Institute of the National Institutes of Health will be asked to do this. The Gallaudet College Public Service Programs Office should then make this

information available to schools for the deaf.

4. Public Service Programs of Gallaudet College should apply for a grant which would fund training programs at schools for paraprofessionals to administer accurate screening tests for retinitis pigmentosa at schools for the deaf and the blind.

5. State vocational rehabilitation offices should include screening for retinitis pigmentosa in their regular medical evaluations for all deaf or blind clients. This will be proposed to the Office of Deafness and Communicative Disorders for possible inclusion in their model statewide plan for services to deaf people.

6. Teacher training programs should include sufficient emphasis on visual problems of deaf children in order to sensitize teachers to the possible presence of visual problems in their students. The Council on Education of the Deaf should explore the feasibility of including at least minimal knowledge of visual problems in their certification requirements.

7. To facilitate referrals for comprehensive testing follow-up evaluations, the National Institutes of Health should compile a list of hospital facilities possessing electroretinogram equipment. Public Service Programs of Gallaudet College will make this information

available to schools.

Counseling and Related Needs

8. The availability of genetic counseling for deaf people must be expanded. Genetic counseling should be based on accurate printed guidelines from specialists in genetics who have an interest in deafness. Dr. Walter Nance will be able to assist with this task.

9. The National Center for Deaf-Blind Youths and Adults should maintain and publish annually occupational information on deaf-blind citizens in order to monitor vocational training and placement. Public Service Programs will bring this to the attention of the appropriate agency.

10. Curriculum and specific program needs such as cane travel, mobility problems and braille instruction need to be determined and appropriate recommendations made to schools for the deaf and for the blind. The Gallaudet College Graduate School will be asked to pursue this matter in connection with their curriculum development for a program in training teachers of multiply handicapped deaf children and youth.

11. The Media Services and Captioned Films Office at the Department of Health, Education, and Welfare should fund development of

instructional materials specific to deaf children with severe visual problems. This recommendation will be forwarded to the Media Services-Captioned Films Office by Public Service Programs.

Public Awareness Needs

12. Papers by deaf-blind adults should be solicited in order to assist others in understanding the life style, potentials, and problems of deaf-blind persons. Dr. Robert Smithdas will be asked to co-ordinate this effort.

13. Dr. McCay Vernon will contact the editors of various professional journals in the area of deafness, such as the Journal of Rehabilitation of the Deaf and the American Annals of the Deaf, to encourage inclusion of articles describing Usher's syndrome and techniques of screening for visual impairments in their publications.

14. Mr. Joe Shinpaugh will be asked to encourage the Conference of Executives of American Schools for the Deaf and appropriate organizations of the blind to stimulate discussion of deaf-blindness at their meetings and in their professional journals.

SYMPOSIUM ON USHER'S SYNDROME

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